



CYP24A1 gene

cytochrome P450 family 24 subfamily A member 1

Normal Function

The *CYP24A1* gene provides instructions for making an enzyme called 24-hydroxylase. This enzyme helps control the amount of active vitamin D available in the body. When active, vitamin D is involved in maintaining the proper balance of several minerals in the body, including calcium and phosphate, which are essential for the normal formation of bones and teeth. One of vitamin D's major roles is to control the absorption of calcium and phosphate from the intestines into the bloodstream. Vitamin D is also involved in several processes in addition to bone and tooth formation.

The 24-hydroxylase enzyme breaks down the active form of vitamin D, called 1,25-dihydroxyvitamin D₃ or calcitriol, to an inactive form when the vitamin is no longer needed. The enzyme also breaks down 25-hydroxyvitamin D (also known as calcidiol), which is the form of vitamin D that is stored in the body.

Health Conditions Related to Genetic Changes

Idiopathic infantile hypercalcemia

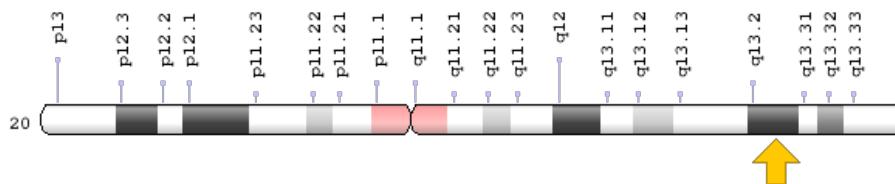
More than 20 mutations in the *CYP24A1* gene have been found to cause a type of idiopathic infantile hypercalcemia called infantile hypercalcemia 1, which is characterized by high levels of calcium in the blood (hypercalcemia) and urine (hypercalciuria) and deposits of calcium in the kidneys (nephrocalcinosis). The hypercalcemia typically causes vomiting, poor feeding, and an inability to grow and gain weight at the expected rate (failure to thrive) in infancy, although some affected individuals do not develop signs and symptoms of the condition until adulthood. Features in affected adults, whether they had symptoms in infancy or not, typically include hypercalciuria, nephrocalcinosis, and kidney stones (nephrolithiasis), although they may not cause any obvious health problems.

The *CYP24A1* gene mutations that cause infantile hypercalcemia 1 reduce or eliminate the activity of the 24-hydroxylase enzyme. A shortage of this enzyme's function impairs the breakdown of calcitriol. The resulting excess of calcitriol increases calcium absorption into the bloodstream, causing hypercalcemia. Dysregulation of calcium absorption in the kidneys leads to hypercalciuria, nephrocalcinosis, and nephrolithiasis.

Chromosomal Location

Cytogenetic Location: 20q13.2, which is the long (q) arm of chromosome 20 at position 13.2

Molecular Location: base pairs 54,145,731 to 54,174,032 on chromosome 20 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 1,25-@dihydroxyvitamin D3 24-hydroxylase
- 1,25-dihydroxyvitamin D(3) 24-hydroxylase, mitochondrial isoform 1 precursor
- 1,25-dihydroxyvitamin D(3) 24-hydroxylase, mitochondrial isoform 2 precursor
- 24-OHase
- CP24
- CYP24
- cytochrome P450 24A1
- cytochrome P450-CC24
- cytochrome P450, family 24, subfamily A, polypeptide 1
- cytochrome P450, subfamily XXIV (vitamin D 24-hydroxylase)
- exo-mitochondrial protein
- HCAI
- HCINF1
- P450-CC24
- vitamin D 24-hydroxylase
- vitamin D(3) 24-hydroxylase

Additional Information & Resources

Educational Resources

- Endotext (2014): Vitamin D
https://www.ncbi.nlm.nih.gov/books/NBK279023/#_ca-phosphate-homeost_toc-vitamin-d_
- Endotext (2017): Vitamin D: Production, Metabolism, and Mechanisms of Action
<https://www.ncbi.nlm.nih.gov/books/NBK278935/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CYP24A1%5BTIAB%5D%29+OR+%28cytochrome+P450+family+24+subfamily+A+member+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- CYTOCHROME P450, FAMILY 24, SUBFAMILY A, POLYPEPTIDE 1
<http://omim.org/entry/126065>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_CYP24A1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=CYP24A1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:2602
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:1591>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/1591>
- UniProt
<https://www.uniprot.org/uniprot/Q07973>

Sources for This Summary

- OMIM: CYTOCHROME P450, FAMILY 24, SUBFAMILY A, POLYPEPTIDE 1
<http://omim.org/entry/126065>
- Dinour D, Beckerman P, Ganon L, Tordjman K, Eisenstein Z, Holtzman EJ. Loss-of-function mutations of CYP24A1, the vitamin D 24-hydroxylase gene, cause long-standing hypercalciuric nephrolithiasis and nephrocalcinosis. *J Urol.* 2013 Aug;190(2):552-7. doi: 10.1016/j.juro.2013.02.3188. Epub 2013 Mar 5.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23470222>
- Molin A, Baudoin R, Kaufmann M, Souberbielle JC, Ryckewaert A, Vantyghem MC, Eckart P, Bacchetta J, Deschenes G, Kesler-Roussey G, Coudray N, Richard N, Wraich M, Bonafiglia Q, Tiulpakov A, Jones G, Kottler ML. CYP24A1 Mutations in a Cohort of Hypercalcemic Patients: Evidence for a Recessive Trait. *J Clin Endocrinol Metab.* 2015 Oct;100(10):E1343-52. doi: 10.1210/jc.2014-4387. Epub 2015 Jul 27.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/26214117>
- Nesterova G, Malicdan MC, Yasuda K, Sakaki T, Vilboux T, Ciccone C, Horst R, Huang Y, Golas G, Introne W, Huizing M, Adams D, Boerkoel CF, Collins MT, Gahl WA. 1,25-(OH)2D-24 Hydroxylase (CYP24A1) Deficiency as a Cause of Nephrolithiasis. *Clin J Am Soc Nephrol.* 2013 Apr;8(4):649-57. doi: 10.2215/CJN.05360512. Epub 2013 Jan 4.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23293122>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3613951/>
- Pronicka E, Ciara E, Halat P, Janiec A, Wójcik M, Rowinska E, Rokicki D, Pludowski P, Wojciechowska E, Wierzbicka A, Ksiazek JB, Jacoszek A, Konrad M, Schlingmann KP, Litwin M. Biallelic mutations in CYP24A1 or SLC34A1 as a cause of infantile idiopathic hypercalcemia (IIH) with vitamin D hypersensitivity: molecular study of 11 historical IIH cases. *J Appl Genet.* 2017 Aug; 58(3):349-353. doi: 10.1007/s13353-017-0397-2. Epub 2017 May 3.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/28470390>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5509812/>
- Schlingmann KP, Kaufmann M, Weber S, Irwin A, Goos C, John U, Misselwitz J, Klaus G, Kuwertz-Bröking E, Fehrenbach H, Wingen AM, Güran T, Hoenderop JG, Bindels RJ, Prosser DE, Jones G, Konrad M. Mutations in CYP24A1 and idiopathic infantile hypercalcemia. *N Engl J Med.* 2011 Aug 4;365(5):410-21. doi: 10.1056/NEJMoa1103864. Epub 2011 Jun 15.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/21675912>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/CYP24A1>

Reviewed: December 2017

Published: June 23, 2020

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services